

# WHAT IS ALPHA-1 ANTITRYPSIN (AAT) DEFICIENCY?



## Could You Have AAT Deficiency?

Sometimes people with AAT deficiency are diagnosed with asthma first. Wheezing is a common symptom for both diseases. Some people with AAT deficiency do not show symptoms at all. People ages 20 to 50 are more likely to have symptoms. Symptoms related to the lungs may include:

- **Shortness of breath or wheezing**
- **Repeated lung infections**
- **Tiring quickly when exercising**

If you have COPD, talk to your doctor about getting tested for AAT deficiency. This is especially important if you have family members with AAT deficiency or who were diagnosed with COPD in their 40s or 50s or have liver disease. A doctor may run a blood test to check your AAT levels. They may also run a genetic test to confirm the blood test results. This is the best way to know for sure if you have AAT deficiency.

Learn the facts about Alpha-1 Antitrypsin (AAT) deficiency, its signs and symptoms, and ways to manage the disease after a diagnosis.

## Basic Facts About AAT Deficiency



**AAT is a protein made in the liver that helps protect the lungs from inflammation caused by infection and inhaled irritants such as cigarette smoke.** Not having enough of this protein can increase the risk of liver and lung diseases.



**AAT deficiency is a disease that runs in families.** The disease is also commonly called “Alpha-1.”



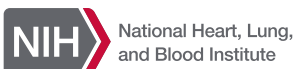
**AAT deficiency can cause chronic obstructive pulmonary disease (COPD).**



**It's important to talk to your doctor about any lung symptoms.** Early diagnosis could help you consider making lifestyle changes and help you be treated with specific therapy for this disease.



**Anyone can have AAT deficiency, regardless of age, race, or ethnicity.**



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### Managing AAT Deficiency

There is no cure for AAT deficiency, but there are ways to slow lung damage.

Augmentation therapy is a lifelong injection treatment that can be used to increase the amount of AAT in your body.

If you develop COPD, you may need medicines to help manage it. Your doctor may also recommend lifestyle changes such as avoiding smoke, dust, and pollution, and quitting smoking.

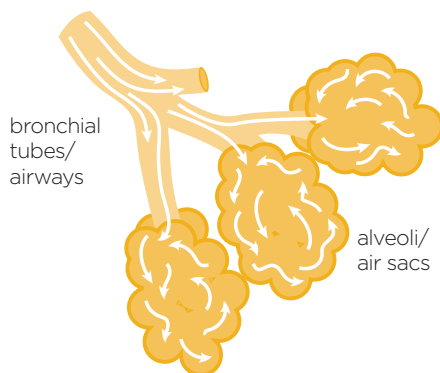
### How AAT Deficiency Affects Your Breathing

When you breathe in, your lungs expand, allowing air to enter your mouth or nose. This air travels to your lungs. AAT protects the lungs from any harmful substances that enter the body.

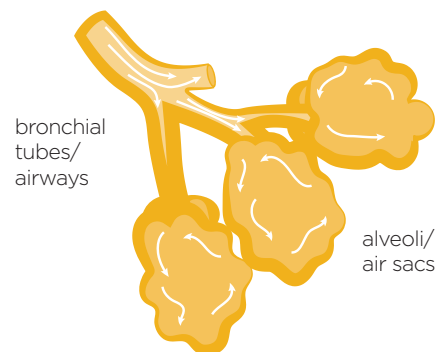
With AAT deficiency, the lungs are more sensitive to harmful substances like dust. Exposure to harmful substances increases damage to the lungs and airways. This can cause shortness of breath, wheezing, or ongoing cough. AAT deficiency can lead to COPD. Less air flows in and out of the airways. The airways can be less elastic. The walls between air sacs can be destroyed and the walls of the airways can be thick and inflamed. The images below show the airways and air sacs of healthy lungs, as well as those of lungs affected by COPD.

### AAT Deficiency Can Cause COPD

#### Breathing with Healthy Lungs



#### Breathing with COPD



For more information, visit [www.nhlbi.nih.gov/health/alpha-1-antitrypsin-deficiency](http://www.nhlbi.nih.gov/health/alpha-1-antitrypsin-deficiency)